

PYRUVATE KINASE DEFICIENCY

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PYRUVATE KINASE DEFICIENCY RELEVANT INFORMATION

- **glycolysis** is a pathway in all cells that metabolizes glucose to pyruvate and produces limited amounts of usable energy as ATP, without dependence on oxygen (Figure 1)
- ATP drives a variety of processes in cells including the sodium-potassium pump that maintains the amount of potassium in cells (Figure 1)
- red blood cells lack mitochondria, the energy-producing compartment found in most cells, and hence can only derive energy from **glycolysis**
- **pyruvate kinase** is the last enzymatic reaction in glycolysis and when deficient in the red blood cell, prevents the cell from producing sufficient amounts of ATP for cell survival, in large part due to the loss of potassium (Figure 1)
- the defective red cells are destroyed in the spleen causing anemia and the spleen enlarges (**splenomegaly**) because it is overworked
- the excessive destruction of red blood cells (**hemolysis**) results in the breakdown of hemoglobin stored in these cells
 - ✓ a by product of hemoglobin breakdown is bilirubin, which when overproduced causes a yellowing of the skin and sclera of the eyes (**jaundice**)
 - ✓ liver processes bilirubin for excretion and the excessive excretion causes inflammation of the gallbladder (**cholecystitis**) leading to formation of bilirubin-containing gallstones (**cholelithiasis**) (Figure 2)

